

Genetic Services

Guideline: Each person should have the opportunity to access services for genetic evaluation. Once acceptance of genetic services has been documented, a genetic service coordinator will coordinate and schedule a genetic evaluation.

DEFINITIONS:

Individual's file: A permanent legal document that provides comprehensive information about the individual's health care status.

Genetic Service Coordinator: An employee of the Greenwood Genetic Center who is responsible for coordinating genetic services for all DDSN individuals in a certain region. The genetic service coordinator is usually a registered nurse, Master's level genetic counselor, or social worker but may be from another discipline. The genetic service coordinator serves as the primary contact regarding genetic services in each region.

Genetic Evaluation: Includes but is not limited to a review of medical and social records, a family interview, a genetic physical examination, appropriate photographic documentation, ordering and obtaining genetic laboratory tests, interpreting laboratory tests, and genetic counseling with the family.

Genetic Services: Services that are a part of the total health care program provided by SCDDSN through a contractual agreement with the Greenwood Genetic Center. Services include genetic evaluation, counseling, and ongoing follow-up.

Geneticist: A physician board certified in clinical genetics who provides comprehensive services to individuals as part of their total health care program.

Primary care providers: Physicians, nurse practitioners, and physician's assistants who provide primary care services and are authorized to prescribe medications and treatments for people on their assigned caseloads.

RATIONALE:

Genetic evaluation is an integral component of the medical services provided by SCDDSN. It can provide valuable information to individuals and their families about the origin and nature of the disability, complications related to the disability, and possible reproductive implications for the person and his or her family. It can also suggest options for future service and treatment needs. Genetic services are a major component of the SCDDSN's prevention efforts.

EXPECTED OUTCOMES:

The goal of genetic services is to determine, where possible, the cause of the presenting disability. If the cause of the disability is determined, the following additional services can be offered:

1. Definition of the risk of a recurrence of the problem in other family members and provide appropriate counseling.
2. Consultation by genetic specialists to the individual's primary care provider and other health care providers as appropriate regarding the management of the particular disability.

EXPECTED OUTCOMES cont'd

3. Assisting the family in connecting with local, state, and national support groups that may provide educational and emotional assistance.
4. Database tracking of the individual to facilitate communication in an efficient manner if and when helpful advances in testing and/or management of their particular disability take place.
5. Periodic re-evaluation of the diagnosed individual provides the opportunity to provide clinical updates including potential future treatment modalities and to answer questions that might have arisen since the initial evaluation.

GENERAL GUIDELINES

1. Each person receiving services by DDSN has the opportunity to receive genetic services.
 - a. Prior to the annual planning meeting the individual's record should be reviewed to determine if the person is receiving genetic services. If there is no signed genetic services consent form in the individual's file indicating that the person or family has accepted or declined the offer for genetic services, a genetic services consent form should be completed at the next scheduled planning meeting.
 - b. Individuals entering community ICF/MR residences from the community should be offered genetic services at the time of admission.
 - c. The original signed genetic services consent form should be retained in the individual's file with a copy sent to the Greenwood Genetic Center Genetic Service Coordinator assigned to that particular region for follow-up as indicated.
 - d. If consent for genetic services is obtained, the assigned genetic service coordinator will coordinate and schedule a genetic evaluation.
 - e. If on the initial evaluation a diagnosis is not apparent, the individual should be scheduled for a re-evaluation in a few years utilizing the most up-to-date diagnostic techniques and tests. Diagnostic capabilities are continually expanding and improving and these return appointments may prove beneficial.
 - f. If genetic services are declined, the services should be offered again at the next annual planning meeting.
2. Reports of the genetic evaluation and recommendations for tests or treatment are sent by Greenwood Genetic Center staff to the regional center/ residential facility.
 - a. Results of the genetic evaluation should be reviewed by the primary care provider and maintained in the individual's record.
 - b. In the event that recommendations made as a result of the genetic evaluation are not followed, the primary care provider should document the rationale for the decision in the medical progress notes.
3. The genetic service coordinator and the geneticist will provide information to the family about the results of the genetic evaluation and recommendations for follow-up.